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Letter to the Editor

Paired Melanotic and Achromic Macules in a Case of Phacomatosis Pigmentovascularis: A Further Example of Twin Spotting?

To the Editor:

Phacomatosis pigmentovascularis (PPV) comprises a telangiectatic nevus and a flat pigmentary nevus arranged in more or less extensive patches. To explain this phenotype, the concept of nonallelic twin spotting was proposed [Happle and Steijlen, 1989]. Twin spots are paired patches of mutant tissue that differ from each other and from the background tissue [Whitehouse, 1982].

In an organism heterozygous for two different recessive mutations located on the same chromosome, the event of somatic recombination may result in two homozygous daughter cells, giving rise to different mutant spots [Stern, 1936; Harrison and Carpenter, 1977; Happle et al., 1990]. According to this concept, PPV includes a third birthmark in the form of nevus anemicus [Ortonne et al., 1978; Hasegawa and Yasuhara, 1985; Hidano and Arai, 1987]. A nevus anemicus can be regarded as a minus counterpart due to allelism at the locus responsible for the pigmentary nevus. Here we report on an unusual case suggesting twin spotting due to allelism at this locus.

An 8-year-old girl was referred to our Department of Dermatology because of cutaneous vascular and pigmentary abnormalities. She had suffered from recurrent episodes of bronchopneumonia since birth. The cutaneous anomalies had first been noted in early childhood. No other relatives had similar lesions.

Physical examination showed a telangiectatic nevus involving her right cheek, as well as a light brown patch localized partly on the right cheek and extending to the right side of the neck (Figs. 1, 2). In addition, a triangular hypopigmented patch was present on the right side of the upper back (Fig. 3). On diascopic pressure, the lesion could be distinguished from the blanched surrounding skin, excluding the differential diagnosis of nevus anemicus. Further checkup, including neurological and ophthalmological examination, did not show any additional anomalies.

Fig. 1. Nevus flammeus on right cheek, extending to upper lip and subpalpebral region.

This unusual case can be taken as yet another clue in the understanding of the origin of PPV. If it is true that the two vascular components observed in this phenotype are caused by allelic mutation [Happle, 1991], one may infer that a similar phenomenon should sometimes occur at the neighboring locus responsible for the pigmentary component. In this way, the present case would represent a "missing link" in our line of thought, explaining PPV as a twin spot phenomenon.

The concept of twin spotting as a genetic basis of PPV

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Fig. 2. Hyperpigmented patch on laterocervical region.

implies the following important aspects. First, PPV would herald the fact that different gene loci controlling either cutaneous vascularization or pigmentation neighbor each other on the same chromosome. Second, the vascular component of PPV may manifest itself as paired patches of telangiectatic nevus and nevus anemicus, reflecting allelism of the underlying genes. Third, the pigmentary component of PPV may manifest itself as paired patches of hyperpigmented and depigmented nevus, reflecting allelism of the underlying genes situated on a neighboring locus.

Theoretically, it is conceivable that a case showing the four components of PPV in the form of both vascular and pigmentary plus/minus nevi will be described in the future. Such cases, however, should be extremely rare, because this would imply compound heterozygosity at two different loci on the same chromosome.

Future molecular research should show whether the proposed concept of both nonallelic and allelic twin spotting in PPV holds true.

Fig. 3. Hypopigmented triangular patch on the back.

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